GNS gene

Glucosamine (N-acetyl)-6-sulfatase

Normal Function

The GNS gene provides instructions for producing an enzyme called N-acetylglucosamine-6-sulfatase. This enzyme is located in lysosomes, compartments within cells that digest and recycle different types of molecules. N-acetylglucosamine-6-sulfatase is involved in the step-wise breakdown of large molecules called glycosaminoglycans (GAGs). GAGs are composed of sugar molecules that are linked together to form a long string. To break down these large molecules, individual sugars are removed one at a time from one end of the molecule. N-acetylglucosamine-6-sulfatase removes a chemical group known as a sulfate from a subset of GAGs called heparan sulfate when the sugar N-acetylglucosamine-6-sulfate is located at the end.

Health Conditions Related to Genetic Changes

Mucopolysaccharidosis type III

Mutations in the GNS gene cause mucopolysaccharidosis type IIID (MPS IIID). Most of these mutations change single DNA building blocks (nucleotides) in the gene. All of the mutations that cause MPS IIID reduce or eliminate the function of N-acetylglucosamine-6-sulfatase.

The lack of N-acetylglucosamine-6-sulfatase activity disrupts the breakdown of heparan sulfate. As a result, partially broken down GAGs accumulate within lysosomes. Researchers believe that the accumulation of GAGs interferes with the functions of other proteins inside the lysosomes and disrupts the normal functions of cells. It is unknown why the buildup of heparan sulfate mostly affects the central nervous system in MPS IIID.
Chromosomal Location

Cytogenetic Location: 12q14.3, which is the long (q) arm of chromosome 12 at position 14.3

Molecular Location: base pairs 64,713,449 to 64,759,406 on chromosome 12 (Homo sapiens Updated Annotation Release 109.20191205, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

• G6S
• glucosamine-6-sulfatase
• GNS_HUMAN
• MGC21274
• N-acetylglucosamine-6-sulfatase
• N-acetylglucosamine-6-sulfatase precursor

Additional Information & Resources

Educational Resources

• Eurekah Bioscience Collection: Defects in Glycosaminoglycan Degradation (Mucopolysaccharidoses)
  https://www.ncbi.nlm.nih.gov/books/NBK6177/#A53462

Clinical Information from GeneReviews

• Mucopolysaccharidosis Type III
  https://www.ncbi.nlm.nih.gov/books/NBK546574
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28GNS%5BTIAB%5D%29+OR+%28glucosamine-6-sulfatase%5BTIAB%5D%29+AND+%28mucopolysaccharidosis%5BTIAB%5D%29%29+OR+%28%28N-acetylglucosamine-6-sulfatase%5BTIAB%5D%29+OR+%28glucosamine-6-sulfatase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+NOT+%28%28Hedgehog%29+OR+%28coevolution%5BTIAB%5D%29%29+AND+english%5Bla%5D

Catalog of Genes and Diseases from OMIM

- N-ACETYLGLUCOSAMINE-6-SULFATASE
  http://omim.org/entry/607664

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_GNS.html

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=GNS%5Bgene%5D

- HGNC Gene Symbol Report

- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:2799

- NCBI Gene

- UniProt
  https://www.uniprot.org/uniprot/P15586

Sources for This Summary


- OMIM: N-ACETYLGLUCOSAMINE-6-SULFATASE
  http://omim.org/entry/607664


Reprinted from Genetics Home Reference:

Reviewed: August 2010
Published: February 11, 2020

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services